A novel NIR spectroscopic qualitative method to monitor the diet compliance in PKU patients.

POSTER n°69 - Theme 3: Share, Care, Rare: Transforming care for rare diseases by 2030


1 Department of Histology, University “Magna Graecia”, Catanzaro, Italy. 2 Department of Medical and Surgical Science, Pediatric Unit, University “Magna Graecia”, Catanzaro, Italy. 3 Centro Regionale di Screening Neonatale, A.O.U. “Mater Domini”, Catanzaro, Italy. 4 Department of Health Sciences, Nephrology Unit, “Magna Graecia” University, Catanzaro, Italy. 5 Ente Ospedaliero Cantonale del Ticino, Istituto pediatrico della Svizzera italiana, Lugano, Switzerland. 6 “Caravaggio” medical center, Toxicology Unit, Roma, Italy. 7 Department of Health Sciences, University “Magna Graecia”, Catanzaro, Italy. *Presenting author

Introduction
Phenylketonuria (PKU MIM 261600) is a human metabolic disease, caused by mutations in the phenylalanine hydroxylase gene. Children born with PKU, thanks to the newborn screening and early dietary interventions, can have relatively normal lives. The diet allows them to keep Phe level in range. The best indicator of dietary compliance is regular monitoring of Phe concentrations by using a Liquid Chromatography (LC) based procedure. We developed a qualitative chemometric model able to separate PKU patients based on their response to the diet in three different categories: Low (<2 mg/dl), Compensate (2,5-6 mg/dl) and High (6,5-20 mg/dl) with NIR spectroscopy.

Methods and results
A qualitative chemometric model was elaborated using 3000 NIR spectra, from 562 DBS PKU samples from the three different groups, based on their Phe levels assessed by LC. Using a sensor calibrated in the Near infra-Red spectrum, 20 scans per sample in duplicate were acquired. Principal component analysis (PCA) was conducted. The chemometric model was validated by blind external procedure. Our model is able to correctly assign unknown sample to the Low, Compensate, or High group with a linearity of 0,91 and a σ=0,078, showing a sensitivity ratio between the number of the correctly classified samples (CCS) and the number of the known samples belonging to each class (KS): Sn=CCS/100KS close to 100%.

Conclusions
We demonstrated that it is possible to use NIR spectroscopy to quickly monitor the diet compliance in PKU patients.